



ZAP70 gene

zeta chain of T-cell receptor associated protein kinase 70

Normal Function

The *ZAP70* gene provides instructions for making a protein called zeta-chain-associated protein kinase. This protein is part of a signaling pathway that directs the development of and turns on (activates) immune system cells called T cells. T cells identify foreign substances and defend the body against infection.

The *ZAP70* gene is important for the development and function of several types of T cells. These include cytotoxic T cells (CD8+ T cells), whose functions include destroying cells infected by viruses. The *ZAP70* gene is also involved in the activation of helper T cells (CD4+ T cells). These cells direct and assist the functions of the immune system by influencing the activities of other immune system cells.

Health Conditions Related to Genetic Changes

ZAP70-related severe combined immunodeficiency

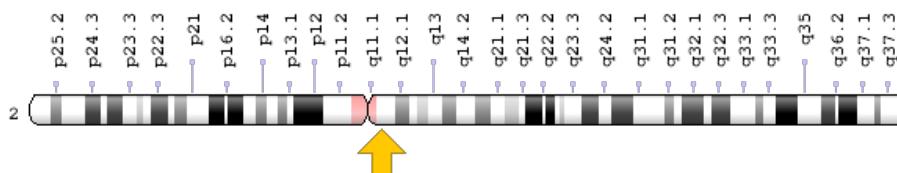
More than 12 mutations in the *ZAP70* gene have been identified in people with ZAP70-related severe combined immunodeficiency (SCID). These mutations either change single protein building blocks (amino acids) in the protein sequence or disrupt how genetic information is pieced together to make the blueprint for producing the protein.

Mutations in the *ZAP70* gene prevent the production of zeta-chain-associated protein kinase or result in a protein that is unstable and cannot perform its function. A loss of functional zeta-chain-associated protein kinase leads to the absence of CD8+ T cells and an excess of inactive CD4+ T cells. The resulting shortage of active T cells causes people with *ZAP70*-related SCID to be more susceptible to infection.

Chromosomal Location

Cytogenetic Location: 2q11.2, which is the long (q) arm of chromosome 2 at position 11.2

Molecular Location: base pairs 97,712,030 to 97,744,327 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FLJ17670
- FLJ17679
- SRK
- STD
- syk-related tyrosine kinase
- TZK
- ZAP-70
- ZAP70_HUMAN
- zeta-chain (TCR) associated protein kinase 70kDa
- zeta-chain associated protein kinase 70kDa
- zeta-chain associated protein kinase, 70kD
- zeta chain of T cell receptor associated protein kinase 70
- zeta chain of T cell receptor associated protein kinase 70kDa

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): Defects in T Cell Function Result in Severe Combined Immunodeficiencies
<https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1509>

GeneReviews

- ZAP70-Related Severe Combined Immunodeficiency
<https://www.ncbi.nlm.nih.gov/books/NBK20221>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ZAP70%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- ZETA-CHAIN-ASSOCIATED PROTEIN KINASE
<http://omim.org/entry/176947>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/ZAP70ID197ch2q11.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ZAP70%5Bgene%5D>
- HGNC Gene Family: SH2 domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/741>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12858
- Mutation Registry for ZAP70 Deficiency
<http://structure.bmc.lu.se/idbase/ZAP70base/index.php>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7535>
- UniProt
<http://www.uniprot.org/uniprot/P43403>

Sources for This Summary

- Elder ME. SCID due to ZAP-70 deficiency. J Pediatr Hematol Oncol. 1997 Nov-Dec;19(6):546-50. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/9407944>
- Elder ME. T-cell immunodeficiencies. Pediatr Clin North Am. 2000 Dec;47(6):1253-74. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11130995>
- GeneReview: ZAP70-Related Severe Combined Immunodeficiency
<https://www.ncbi.nlm.nih.gov/books/NBK20221>

- Grunebaum E, Sharfe N, Roifman CM. Human T cell immunodeficiency: when signal transduction goes wrong. *Immunol Res.* 2006;35(1-2):117-26. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/17003514>
- Picard C, Dogniaux S, Chemin K, Maciorowski Z, Lim A, Mazerolles F, Rieux-Lauca F, Stolzenberg MC, Debre M, Magny JP, Le Deist F, Fischer A, Hivroz C. Hypomorphic mutation of ZAP70 in human results in a late onset immunodeficiency and no autoimmunity. *Eur J Immunol.* 2009 Jul; 39(7):1966-76. doi: 10.1002/eji.200939385.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19548248>
- Roifman CM, Dadi H, Somech R, Nahum A, Sharfe N. Characterization of ζ -associated protein, 70 kd (ZAP70)-deficient human lymphocytes. *J Allergy Clin Immunol.* 2010 Dec;126(6):1226-33.e1. doi: 10.1016/j.jaci.2010.07.029.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20864151>
- Turul T, Tezcan I, Artac H, de Bruin-Versteeg S, Barendregt BH, Reisli I, Sanal O, van Dongen JJ, van der Burg M. Clinical heterogeneity can hamper the diagnosis of patients with ZAP70 deficiency. *Eur J Pediatr.* 2009 Jan;168(1):87-93. doi: 10.1007/s00431-008-0718-x. Epub 2008 May 29.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18509675>
- OMIM: ZETA-CHAIN-ASSOCIATED PROTEIN KINASE
<http://omim.org/entry/176947>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/ZAP70>

Reviewed: November 2009

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services